KENTUCKY NEWBORN SCREENING PROGRAM

PARENT TEACHING SHEET

Kentucky Newborn Screening Program, 275 East Main Street, Frankfort, KY 40621, Phone (502) 564-3756, Fax (502) 564-1510

Hyperphenylalaninemia (H-PHE)

CAUSE

PKU is one type of amino acid disorder. People with PKU have trouble breaking down an amino acid called phenylalanine from the food they eat. PKU occurs when an enzyme called "phenylalanine hydroxylase" is either missing or not working properly. This enzyme's job is to break down the amino acid phenylalanine. When a child with PKU eats foods with phenylalanine, it builds up in the blood and causes problems. It is found in almost every food, except pure fat and sugar.

IF NOT TREATED

The symptoms can vary from person to person. Babies with PKU seem perfectly normal at birth. The first effects are usually seen around 6 months of age. Untreated infants may be late learning to sit, crawl, and stand. They may pay less attention to things around them. Without treatment, a child with PKU will have learning or developmental delays.

Some of the effects of untreated PKU are: learning or developmental delays, behavioral problems, hyperactivity, restlessness or irritability, seizures, eczema, musty or mousy body odor, fair hair and skin.

TREATMENT OPTIONS

- Your doctor will work with a metabolic specialist and dietitian to care for your child. Lifelong treatment is usually needed.
- Prompt treatment is needed to prevent learning or developmental delays. Newborns need to drink a special medical formula. It is possible to still breastfeed your baby as long as you get help from a dietitian. Babies who are breastfed usually need the medical formula as well.
- A diet low in phenylalanine (Phe) is generally prescribed. Your child must not have cow's milk, regular formula, meat, fish, eggs or cheese. You must avoid the sugar substitute aspartame (Equal, NutraSweet) because it contains high levels of phenylalanine and can quickly raise the blood levels of Phe in people with PKU.
- Blood levels with have to be checked regularly to monitor the levels of PHE. The diet and formula may have to be adjusted.
- Contact your child's doctor immediately at the start of any illness.

IF TREATED

Children who receive early treatment and keep their Phe levels with the suggested ranges may be able to live healthy lives with typical growth and development. Some children may continue to have learning difficulties and other health problems despite treatment.

If treatment is not started until 6 months of age, learning or developmental delays often occurs. Treatment, even if started late, can help control behavior and mood problems and further prevent further damage to the brain.

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